

WHAT IS CLAIMED IS:

1 1. An isolated nucleic acid and the degenerate
2 sequences thereof, which encodes human $\alpha 1$ chain collagen
3 protein, comprising the nucleotide sequence set forth in SEQ
4 ID NO. 5.

1 2. The nucleic acid as claimed in claim 1, wherein the
2 human $\alpha 1$ chain collagen protein encoded by the nucleic acid
3 has the amino acid sequence set forth in SEQ ID NO. 1.

1 3. The nucleic acid as claimed in claim 2, wherein the
2 human $\alpha 1$ chain collagen protein comprises:

3 (i) von Willebrand factor A domain set forth in SEQ ID
4 NO. 2;

5 (ii) thrombospondin N-terminal-like domain set forth in
6 SEQ ID NO. 3; and

7 (iii) collagenous domain set forth in SEQ ID NO. 4.

8 4. The nucleic acid as claimed in claim 1, comprising
9 DNA and RNA.

1 5. The nucleic acid as claimed in claim 4, wherein the
2 DNA comprises cDNA and genomic DNA.

1 6. A human $\alpha 1$ chain collagen protein having the amino
2 acid sequence set forth in SEQ ID NO. 1.

1 7. The human $\alpha 1$ chain collagen protein as claimed in
2 claim 6, wherein the protein is encoded by the nucleic acid
3 of claim 1.

1 8. The human $\alpha 1$ chain collagen protein as claimed in
2 claim 7, comprising:

3 (i) von Willebrand factor A domain set forth in SEQ ID
4 NO. 2;

5 (ii) thrombospondin N-terminal-like domain set forth in
6 SEQ ID NO. 3; and

7 (iii) collagenous domain set forth in SEQ ID NO. 4.

1 9. A recombinant vector comprising the nucleic acid of
2 claim 1 and a regulatory sequence.

1 10. The recombinant vector as claimed in claim 9,
2 wherein the regulatory sequence comprises an operatively
3 linked promoter.

1 11. The recombinant vector as claimed in claim 9,
2 wherein the recombinant vector is designated Bluescript
3 KS(+)/*E. coli* DH5 α (*hCOLA1*) and deposited at the Culture
4 Collection and Research Center (Hsinchu, Taiwan) and
5 assigned accession number CCRC 940331.

1 12. A method for producing human $\alpha 1$ chain collagen
2 protein, comprising the steps of:

3 (a) transforming or transfecting a host cell with the
4 recombinant vector of claim 9;

5 (b) culturing said transformed or transfected cell
6 under the conditions sufficient for expression of the human
7 $\alpha 1$ chain collagen protein; and

8 (c) recovering and purifying the human $\alpha 1$ chain
9 collagen protein.

1 13. The method as claimed in claim 12, wherein the host
2 cell is selected from the group consisting of prokaryotic
3 and eukaryotic cell.

1 14. The method as claimed in claim 13, wherein the
2 prokaryotic cell comprises *Escherichia coli*.

1 15. The method as claimed in claim 13, wherein the
2 eukaryotic cell comprises mammalian cell.

1 16. The method as claimed in claim 12, wherein the
2 recovering and purifying step is conducted by column
3 chromatography.

1 17. An isolated nucleic acid comprising at least 500
2 contiguous nucleotides in length derived from SEQ ID NO. 5
3 or a complementary nucleotide sequence thereto.

1 18. A kit for detecting the disease related to the
2 mutation of SEQ ID NO. 5 in a mammal or human comprising a
3 probe, which comprises the nucleic acid of claim 1 or claim
4 17.

1 19. An isolated nucleic acid comprising at least 20
2 contiguous nucleotides in length derived from SEQ ID NO. 5
3 or a complementary nucleotide sequence thereto.

1 20. A kit for detecting the disease related to the
2 mutation of SEQ ID NO. 5 in a mammal or human comprising a
3 primer, which comprises the nucleic acid of claim 19.